ARUP LABORATORIES

CYSTIC FIBROSIS (CF) TESTING

descriptions and ordering recommendations

Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants (2013661)

Tests for 165 pathogenic variants.
Order for the following indications:
- Carrier testing in a healthy patient
- Patient has symptoms of CF
- Patient has family history of CF

Cystic Fibrosis (*CFTR*), 165 Pathogenic Variants, Fetal (2013662)
0050608—Maternal sample is recommended to rule out maternal cell contamination.

Tests amniocytes for 165 pathogenic variants.
Order for fetal samples with the following indications:
- Both parents have *CFTR* variants which are known to be included on the panel
- Echogenic or cystic dilatation of the fetal bowel

Cystic Fibrosis (*CFTR*) Sequencing (0051110)

Detects 97 percent of the more than 2,000 described *CFTR* gene variants.
Order for patients with symptoms of classic CF or a CFTR-related disorder but without two variants identified by the CF 165 pathogenic variants.

Cystic Fibrosis (*CFTR*), 165 Variants with Reflex to Sequencing (2013663)

Tests for 165 pathogenic variants. If two *CFTR* variants are not identified, sequencing is performed to detect 97 percent of the more than 2,000 described *CFTR* variants.
Order for patients with the following indications if not previously tested using the CF 165 pathogenic variants test:
- Patients with symptoms of classic CF
- Patients with a positive sweat chloride
- Patients with symptoms of a CFTR-related disorder

Cystic Fibrosis *(CFTR)* Sequencing with Reflex to Deletion/Duplication (0051640)

Tests for more than 2,000 *CFTR* gene variants. If two pathogenic *CFTR* variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.
Order for patients with symptoms of classic CF or a CFTR-related disorder but without two variants identified by the CF 165 pathogenic variants test.

Cystic Fibrosis *(CFTR)* Deletion/Duplication (0051642)	**Tests for large deletions or duplications in the *CFTR* gene, accounting for 1–2 percent of variants.** Order for the following patients: • Symptoms of classic CF or a CFTR-related disorder without two identified pathogenic variants following *CFTR* sequencing. • To determine carrier status in individuals who have a family member with a large *CFTR* gene deletion.
Cystic Fibrosis (*CFTR*), 165 Variants with Reflex to Sequencing and Reflex to Deletion/Duplication (2013664)	**Tests for 165 pathogenic *CFTR* pathogenic variants. If two pathogenic *CFTR* variants are not identified, sequencing is performed. If two *CFTR* variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.** Order for patients with a clinical diagnosis of CF as a cost-effective method for determining the causative variants present.

visit our website for more information:
WWW.ARUPLAB.COM

ARUP is a nonprofit enterprise of the University of Utah and its Department of Pathology.

ARUP LABORATORIES
500 Chipeta Way
Salt Lake City, UT 84108
Phone: (800) 522-2787
Fax: (801) 583-2712

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